sensory neuropathy (HMSN), intellectual disability with neuronal migrations defects and cortical malformations such as posterior predominant pachygyria, lissencephaly and corpus callosum anomalies. We present twin girls with severe congenital cerebral malformations, refractory epilepsy and acquired cataracts due to a de novo mutation in DYNC1H1 identified in girl 1. **Methods:** Description of twin girls with severe cortical malformation and acquired cataract and family based gene sequencing analysis to identify de novo mutation in DYNC1H1 in girl 1. **Results:** After prenatal sonographic detection of corpus callosum anomalies in both twins in an otherwise uneventful pregnancy, postnatal cMRI in girl 1 showed partial agenesis of corpus callosum, bilateral polymicrogyria and asymmetric ventricles; in girl 2 subtotal agenesis of corpus callosum, hypoplastic cerebellum and multiple cortical dysplasias. Both girls developed severe intellectual disability, intractable epilepsy since the age of 4 (girl 1) resp. 6 month (girl 2), spastic (1) resp. ataxic (2) cerebral palsy and microcephaly. Cataracts occurred in both girls at the age of 12 months. A gene panel analysis identified a de novo mutation in the DYNC1H1-Gen (c.11015C>T/p.Ser3672Leu) in girl 1 considered pathogenic. **Conclusion:** Our findings broaden the clinical spectrum and expand the phenotypes associated with DYNC1H1-mutations and suggest a role of dynein in human ocular development. The observation confirms the role of DYNC1H1 in central and peripheral neuronal function and could have implications on molecular diagnostics in complex cortical malformations by adding DYNC1H1 to related genes in microtubule-dependent motor protein pathways.

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P1-146
Cerebral quantitative DTI and tractography in 25 patients with PLP1-related disorders

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Objective: We used brain diffusion tensor imaging (DTI) and tractography to analyze the severity of patients with PLP1-related disorders (Pelizaeus- Merzbacher disease and spastic paraplegia type 2). **Methods:** Twenty-five male patients (ranged from 0.7 to 43.5 years) with PLP1- related disorder were included in this DTI study. Subjects were classified according to best motor function acquired before 5 years into five categories (from PMD0 without motor acquisition to PMD4 with autonomous walking). We performed a quantitative DTI study on different brain regions of white or grey matter, tracking of the cortico-spinal fascicles (CSF) and whole brain tractography. **Results:** No difference was observed between severity groups on quantitative DTI parameters (apparent diffusion coefficient, fractional anisotropy, radial and axial diffusivities) using two different DTI software. Tractography of the CSF did not reveal significant differences that could permit to distinguish severity groups. On the contrary, whole brain tractography seems to be a more relevant approach with high abnormalities in fibers from semi-oval centers and corpus callosum linked to clinical severity. **Conclusion:** To distinguish early the clinical severity of patients with PLP-pathies is a present challenge to consider early therapeutical approaches in these disorders. Tractography of whole brain could be an interesting way of classifying patients and following disease's severity.

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P1-147
Mental retardation among children with cerebral palsy as observed in Nepal with a small trial with nootropic (Modafinil)

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Objective: The scenario of CP in a developing country like Nepal is different from the developed countries. This study describes the association between ID and CP along with different co-morbidities and studies the trial with Modafinil. **Methods:** Between January 2014 to June 2016, 438 children between 3 to 16 years with CP at a rehabilitation center (Self-help Group for Cerebral Palsy, Nepal) were assessed. Kaufman Assessment Battery for Children-II was used to assess their IQ. Type of CP was classified by clinical phenotype, severity by Gross Motor Function Classification System and Quality of Life(QoL) as per Cerebral palsy quality of life questionnaire for children manual. Children with vision impairment were not included in the study. Modafinil was used in 44 children with mild ID for 1 year. **Results:** Among the CP children 48% (n=210) were found to have IQ below average, where 151 children had microcephaly. Intellectual Disability(ID) was more common in CP children with Epilepsy. 31% of dyskinetic, none of ataxic and 63% with spastic CP had ID. QoL was found to be directly proportional to the severity of CP and ID with highest scores on "Social wellbeing and acceptance" and "Emotional wellbeing and self-esteem domain" while lowest points in "Pain and Impact of disability". Neuroimaging did not co-relate with the severity of CP or ID. Modafinil was associated with increase in IQ by a mean of 3-6 points, however, parents reported that children responded better to external stimuli and paid more attention. **Conclusion:** Severity was the determining factor for QoL. Neuroimaging alone was an unreliable predictor of severity of ID or CP. The increase in IQ by modafinil was not significant but may be tried in inattentive or drowsy children. However, more study is required regarding use of modafinil for ID in CP. Also, the cause of high incidence of microcephaly also must be investigated.

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P1-148
PEHO syndrome – The end point of severe epilepsies

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Objective: Progressive encephalopathy, hypsarrhythmia, oedema and optic atrophy (PEHO) is a recognised rare clinically distinct syndrome. Children with this condition have severe epilepsy and the prognosis is poor. Those who do not satisfy all essential criteria are deemed PEHO-like. Our objective was to study the underlying genetic basis for the condition and to gain understanding of processes causing epileptic Encephalopathies (EE). **Methods:** We studied a cohort of 19 children with differing racial backgrounds